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A State-Level Retrospective Analysis of Newborn Screening for Sickle Cell Hemoglobinopathies

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INTRODUCTION

Dr. Robert Guthrie was at the forefront of early Newborn screening in the US in the 1960s. Early detection, diagnosis, and treatment of certain genetic, metabolic, or infectious congenital disorders have led to significant improvement in long term health outcomes.

Newborns affected with Sickle Cell Disease (SCD) will appear healthy at birth, but anemia develops in the first few months of life, followed by increased risks to other of life-threatening conditions. Treatment such as penicillin prophylaxis, proper vaccination, and long-term management have tremendously reduced the complications of SCD.

Nebraska law mandated Newborn screening for the first condition in 1967 and has expanded to 32 conditions. State-mandated newborn screening programs have led to significant advances in the care of infants born with SCD. To further contribute to the progress that has already been made, we aimed to review the outcomes of newborn screening including the incidence of sickle cell hemoglobinopathies and the rate of confirmation testing by primary care providers.

SCD PATHOPHYSIOLOGY

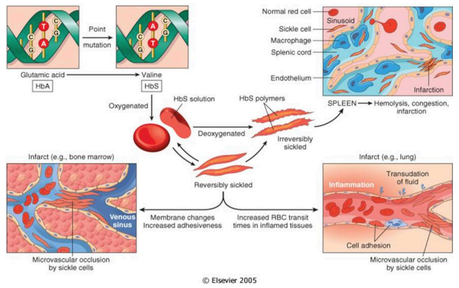
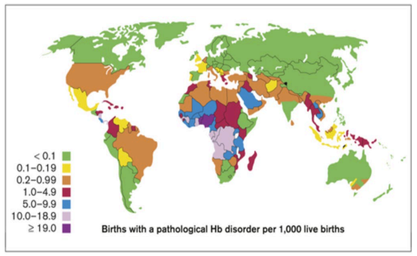


Fig1: Pathophysiology of sickle cell disease (Image from Elsevier 2005, google images)

SCD are inherited abnormalities in the function of hemoglobin.

- Single gene mutations to the formation of abnormal hemoglobin such as Hemoglobin S (HbS), Hemoglobin C (HbC), Hemoglobin D, Hemoglobin FA Barts, etc.
- Single-gene disorders
- Red blood cells ability to carry oxygen from the lungs to other parts of the body is affected
- Over 600 hemoglobin diseases defined by the American College of Medical Genetics.
- Most catastrophic abnormal hemoglobin conditions are sickle cell anemia and sickle beta-thalassemia
- Sickle cell anemia mostly affects people of African descent, but the disease can also affect people of Hispanic, Arabic, Indian or Mediterranean descent



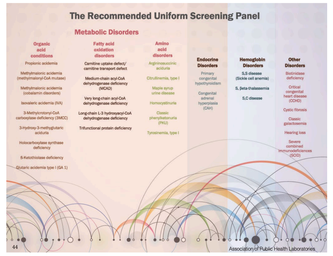


Fig 2a: World map of births with a pathological Hb disorder (WHO, June 2008; Weatherall DJ, Blood 2010)
Fig 2b: Recommended screening panel, association of public health laboratories, pub 2013, aphl.org, web

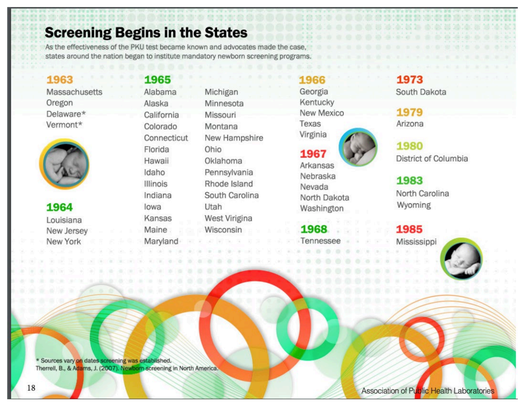


Fig 3: Recommended screening panel, association of public health laboratories, pub 2013, aphl.org, web

METHOD

Deidentified data was obtained from the state of Nebraska Department of Health and Human Services (DHHS) newborn screening program.
Using state-level screening information, the crude incidence rate of sickle cell trait was calculated as the screened positives divided by the total number of screened newborns within the state of Nebraska from 2011 to 2018. Rates were further delineated by mutation type. A Daniel's test was utilized to determine if the crude incidence rates exhibited characteristics of a trend. Rates were also examined with joinpoint regression to determine annual percent change (APC). Excel v 1808 and Joinpoint Trend Analysis v 4.5 software packages were used for all analyses; $p < 0.05$ was considered significant.

RESULTS

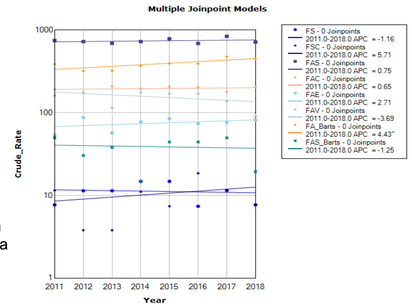


Fig 4: Crude incidence rate of sickle cell trait within the state of Nebraska (2011-20180

CONCLUSION

The data obtained from this study showed that there were no significant changes in the incidence of the SCT in the state of Nebraska from 2011 to 2018. However, there was a significant increase in the incidence of Hemoglobin FA Barts.

Hemoglobin FA Barts is associated with alpha thalassemia which is most prevalent in Southeast Asia but also common in the Mediterranean, Africa, middle east, central Asia and India.

The changing migration pattern in the state of Nebraska could explain the findings noted above. Appropriate patient and family's education are crucial and necessary to improve the long-term outcomes related to these hemoglobinopathies.

For the healthcare system to continue to provide the appropriate care, future studies can aim to further elucidate these trends and patient demographics of this population in Nebraska. In addition, review the process of notification, methods of follow up, as well as describe the resources available to families after receiving a positive screening result.

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